Brief Clinical Report

Trisomy 1 in a Clinically Recognized Pregnancy

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Cytogenetic analysis of products of conception has identified trisomies for all human chromosomes except for chromosome 1. Presented here is a patient with clinical signs of pregnancy, including increasing hCG levels and evidence of a gestational sac on ultrasound. However, the pregnancy was lost between 8–9 weeks post-LMP with the diagnosis of a blighted ovum. Cytogenetic analysis of cultured chorionic villi demonstrated a 47,XY,+1 chromosome complement in all cells. This is the first reported case of nonmosaic trisomy 1 in a clinically recognized human pregnancy. Am. J. Med. Genet. 68:98, 1997 **© 1997 Wiley-Liss, Inc.**

KEY WORDS: trisomy 1; spontaneous abortion; cytogenetic analysis of products of conception

INTRODUCTION

Cytogenetic analysis of products of conception has been performed since the early 1960's, and since that time, trisomies for all human chromosomes except chromosome 1 have been identified in clinically recognized pregnancy loss. Trisomy 1 conceptions are theoretically probable; however, they are likely lost before the first missed menstrual period, and as such, are not ascertained. We report on the first case of a clinically recognized pregnancy with nonmosaic trisomy 1.

CLINICAL REPORT

The patient (G2P0) presented for obstetric care at 7.5 weeks post-LMP (last menstrual period). hCG levels were measured starting at 6 weeks of gestation and were increasing, although not as well as expected. hCG levels were as follows: 6 weeks, 447 mIU/ml; 6.2 weeks, 707 mIU/ml; 7 weeks, 1,102 mIU/ml; 7.5 weeks, 1,500 mIU/ml; 8 weeks, 1,626 mIU/ml; 9 weeks (post D & C), 222 mIU/ml; and 10 weeks, 17 mIU/ml.

Her first ultrasound (abdominal) study was performed at 7.5 weeks of gestation and showed the suggestion of an intrauterine sac, .9 × .6 × .8 cm, with no fetal pole or free fluid seen. A second (transvaginal) scan, performed 5 days later, showed what appeared to be a deteriorating sac, .5 × .6 × .6 cm, isolated in the endometrium. At 8.5 weeks of gestation, vaginal bleeding ensued, and no evidence of a gestational sac was found on ultrasound. A D & C was recommended and performed; diagnosis was blighted ovum.

Cytogenetic analysis from dissected chorionic villi (cultured cells, no direct preparation) demonstrated a 47,XY,+1 chromosome complement. There was no possibility of this being due to cultural artifact, as this result reflected analysis of cells from multiple cultures.

CONCLUSIONS

This case documents the first known example of a clinically recognized pregnancy with trisomy 1. There has been one previous report of trisomy 1, in an eight-cell human preembryo formed via in vitro fertilization [Watt et al., 1987]. In the case reported here, implantation and formation of a fetal sac were documented by ultrasonography although no fetal development appeared to have taken place as no fetal pole was ever visualized. Based on this, therefore, a trisomy 1 conception is capable of early cell cleavage and implantation, but it is not compatible with fetal development to any degree.

REFERENCES

Watt JL, Templeton AA, Messinis I, Bell L, Cunningham P, Duncan RO (1987): Trisomy 1 in an eight cell human pre-embryo. J Med Genet 24:60–64.

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Received 11 March 1996; Accepted 29 April 1996